

Figure 1

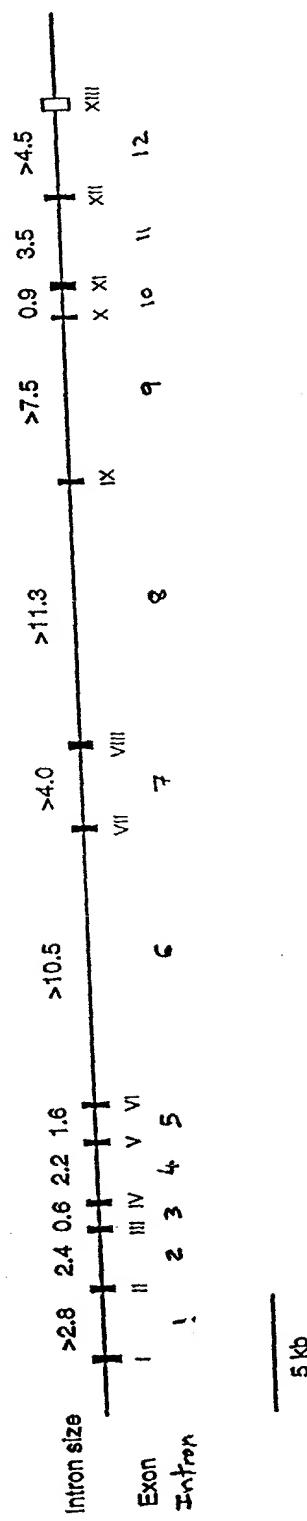


Figure 2A

promoter and exon 1

ACTGCGGAGATGAGGGTCTAGAAAGGTGGTGGCGGGCAT  
GTGGACCGTTGTAAGGGCTCTGGGTTCCCTGGGTGGCCT  
**GGCGAAGT**CTACTCACAGTGACCAACCATGATGATGGT  
CCCGATAGAGGAGGAGAGGGAGGGAGGGAAAAGGAAG  
GGTAGAGGGCTCAGAGGGAGAGCTGGGAGGAGGGAGA  
CATAGGTGGGGAAGGGTAGGAGAAAGGGGAAGGGAGC  
AAGAGGGTGAGGGCACCAAGCCCCATAGACGTTTGGC  
TCAGCGGCCACGAGGCTCATCAGCTCCCCCCCCAAC  
GGAAGCGAGGCCGTGGGGCAGCGGCAGCATGGCGGGC  
TTGTCTTGGCGGCCATGCCCGCCCCCTGCCCGTCCGA  
TCAGCGCCCCCGCCCGTCCCCGCCCGACCCCGCCCCCG  
GCCCGCTCAGGCCCGCCCTGCCGCCGGAAATCCTGAAG  
CCCAAGGCTGCCGGGGCGGTCCGGCGGCCGGCGAT  
**GGGGATAAAACCACTGCCACCTGCCGGCTGCTCC**

TGCGTGCCTGCCGTCCCGATCCACCGTGCCTCTGCGG  
CCTGCGTGCCTGGAGTCCCCGCCTGTGTCGTCTCTGTCG  
CCGTCCCCGTCTCCTGCCAGGCGCGAGACATGGCTGCTCCGC  
GCGGTGGGCCAGGCGCGAGACATGGCTGCTCCGC  
CAAAGCGCTGGCTGCCGGCGCTGGCGTGTGATGATGAT  
GCTACTGTGCGCTGTGCTGGCGCTGTGATGATGATGAT  
GGTGCCGTGCTCATCAAGCAGCAGGTCTTAAG

A

GTGGGTGAGGGAGACCCAGGGGTCCGCCACGGACCC  
GGGCTGTTGGCGCTGGCGCCGGAGGACCCGCGCGTT  
GCGGTGGGTGGCGACCGCAGCGGAATCGCGCCCGGGC  
CTGGCGCCGAGAACACGAGGGAGGCCAGGCGCTTCCGG  
AGGGGCTGCTGCCCGCCTCCCCACCAACCTCACC

## Figure 2B

### exon 2

AGCCTCATGTGCGAAGGGTTCCACCACCTCCTATCC  
CAAGCTCCC GCCGAGGAGCCCTTCCCTGGCCGGGCTCG  
GGCAGCTGTTCCGGAGCCTGTGGTGGGGCGTGGGCC  
CTCATCACTCTCCTCACAGCGTACTTGTCCCTCCCC  
CTGCAG

AACGTGCGCATCGACCCCCAGTAGCCTGTCCTTCAACATG  
TGGAAGGAGATCCCTATCCCCCTCTATCTCTCCGTCTAC  
TTCTTGACGT CATGAACCCCCAGCGAGATCCTGAAGGGC  
GAGAACGCCAGGTGCGGGAGCGCGGGCCCTACGTGTAC  
AG

GTGAGGGCTGTGTCACCGTGTGGTGGACGGGCCGGCTGA  
CGCTGGCATGGGACGGGCTCANAGTGGACGGGATG  
GGGAGGGCTGCTGACTGACCCCCAAACATTGTTCCGGAA  
GCACGCAACTCATAGTCGGGTAAGTGTACTCCCCAAA  
AAGTTTGCCT

### exon 3

CATGTCTGCAGTGGCAGGCAGCGGGAGGGACAGACTT  
GGCGAAGGGGCCGAGCTCAGCTTGGCTGTGGGCCCGGA  
GGTGTGCACAGACGTCCAGGGCCCTGGTCCAGGCAG  
GCATTGCAGGCAGTAGAAGGGAAACGTCCCAGCAG  
CGGGGCGGGCGTGTGACCCACTGGCTCCCCACAG

GGAGTT CAGGCACAAAAGCAACATCACCTTCAACAACAA  
CGACACCGTGTCTTCCCTGAGTACCGCACCTCCAGTT  
CCAGCCCTCCAAGTCCCACGGCTCGGAGAGCGACTACAT  
CGTCATGCCAACATCCTGGTCTTG

<sup>A</sup>  
GTGAGGGCTGCCCTGTGGCCCACGCCGCCCTCGCACCCCTGA  
CCTCGTCCCCCTGTCTCTCCCTCCGCCCTGCCCTGTG  
CAGAGAGCAGTCCCTGAGGTGGTGGAGCGTGGGGACTC  
ACGCCCTGGTGGTGGCTTCCGCCCTGTGCTGTCTCCAC  
CACCCCCA

## Figure 2C

### exon 4

GGTGGTTCTGGTGTCCCAGATGCCCAACGTGGCCACTCC  
AGGGGCCTCCTGCACCCCAGCATTCCTCATGGGCT  
CTTTGCTGTGAGGCCAGCTGGGCAAGGGAGGATG  
GGCCAGCCACGTCCAGCCTCTGACACTAGTGTCCCTCG  
CCTTGCA

GGTGCGGCGGTGATGATGGAGAATAAGCCATGACCCTG  
AAGCTCATCATGACCTTGGCATTCAACCACCTCGCGAA  
CGTGCCTTCATGAACCGCACTGTGGGTGAGATCATGTGG  
GGCTACAAGGACCCCTTGTGAATCTCATCAACAAGTACT  
TTCCAGGCATGTTCCCTCAAGGACAAGTTCGGATTAT  
TTGCTGAG

GTACGTGTGGCCTGGTGAGAAGCCAAGATTCAAGGCCTG  
TGTCCCTGTCTTCCCCTCACACAGCCTGGACACTGGTC  
ACCAGCTTGCTTGTAGCTGGCTGGGATCTAGTGGCTG  
TGGGTGTAAAGTGAUTGAGAACCTGACTCAAACCGGCTT  
GAGTGAAA

### exon 5

CCTCTCGGTCCCCAGACACTGGGCATTGGCAGTGAACC  
AGATGCTGGGGGCCCTGTCTTCTGGTGGAGGGGAGGA  
GGGCTCAGCCCAGAATGTTCAAGACCAGGCCGGCTCAA  
TGGCAGGCCTAACGCTTACGATGCTGTTCCCTGCTGTGT  
CTGTAG

CTCAACAACCTCCGACTCTGGCTCTCACGGTGTTACG  
GGGGTCCAGAACATCAGCAGGATCCACCTCGTGGACAAG  
TGGAACGGGCTGAGCAAG

GTGAGGGCGAGAGGGGAGGGCCCTGTGCCAGGGAGA  
GGGGAGGGTGGGCCGGGCATGGCTGCTGGGAGTGGCA  
GGGACCAAGAGAGCTCCTCTTGTGCTGAAGAG  
GGTGCCTGGAGGATGAACACTCTGAAGTGGAGGAGGG  
ATTTTA

## Figure 2D

exon 6

TCTCTGTGTCTACATAGCCTGCCCTTCCCACCGTG  
CCAGTATTGGGAATTGAGTGGCCGTGCACCAAGGGT  
GAGTTAGGTGTGCAGCACCTGAGAGGGCTTATTAAGG  
GGCCTTGGCCCTACTGAGGGTCTAGTCTGGATGCTTCC  
CCCCAG

GTTGACTTCTGGCATTCCGATCAGTGCAACATGATCAAT  
GGAACTTCTGGCAAATGTGGCCGCCCTCATGACTCCT  
GAGTCCTCGCTGGAGTTCTACAGCCGGAGGCCTGCCG

GTAATCACTGGGACTCGGGGCCTCCTGGTTTCCTGGT  
AGCTCATGGCCAATTCTGTGGTGTGGCTGTGCACTT  
GGAAAGCATTGACTCATCGTGGATTGACTCAGTAG  
CCCTGGCACCAAGCTTGAATTCTCTGGTCACACCACC  
AAAAGC

exon 7

GGAGGTCGCTGCAGCTCCGGGTGAGAGATGGGGCGG  
TTTGGACCCGGGAGGTGGTAGCGCCGTGGGAGAAGTG  
GCTGGATCTGGCAGCCTTGGCAGGGCTGGCTGGC  
CGCCGGGTCTGGGTGTCCCCTCTCATCCTGTCTGTCC  
CCTGCAG

ATCCATGAAGCTAATGTACAAGGAGTCAGGGGTGTTGA  
AGGCATCCCCACCTATCGTTCTGGCTCCAAAACCT  
GTTTGCCAACGGTCCATCTACCCACCCAACGAAGGCTT  
CTGCCCGTGCCTGGAGTCAGGAATTCAAGACGTACAC  
CTGCAGGTTCA

GTACGTGCCGTCCCTGTTCTGGGATNGCCGGAGGGTGT  
TAGGTNTNGGCACCTNANGTTATCTGCCAATGCTG  
TCTGCTTAATCTCTGGCCTCTGTACTCTTGATAACC  
CATTAAGCCAAAATATGATGCCTCTGGGACGATATCTG

## Figure 2E

exon 8

TGGGGCTTTTACAGAATGGAGGAAGGGATCCTCTCT  
GTCGGGTATTATGGTCATGCCACGGGGTGCCTGCAG  
ACCACAGCTCTGTGCAGACTCCGGAGTGGCAGGACGTG  
CCAATATACTGTCGTGTATGATGTCCCCCTCCCTGCCCT  
TGGTAG

GTGCCCCCTGTTCTCTCCCATCCTCACTTCCTCAACG  
CGAACCGGTTCTGGCAGAAGCGGTGACTGGCCTGCACC  
CTAACCAAGGAGGCACACTCCTTGTCCTGGACATCCACC  
CG

GTGAGCCCCCTGCCATCCTCTGTGGGGGTGGTGATTCC  
TGGTTGGAGCACACCTGGCTGCCTCTCTCCCCAG  
GCAGAGAGCTGCTGTGGCTGGGTGGTGGGAAGCCTGG  
CTTCTAGAATCTCGAGCCACCAAAGTCCCTACT

T

exon 9

CCCCAGCCCTGCGCTTGTAGGTAAGATAACAAGCAAG  
CTCCACTGGCAGTAGCTGGACGCCACCCCTTGAC  
TGGGACCAAGGAAAAGAAGGTTGACTGTGTCCCTGGA  
GCTTGGGGGTGCCAGTCTCACTGTGTTGTCGCG  
CAG

GTCACGGGAATCCCCATGAACTGCTCTGTGAAACTGCAG  
CTGAGCCTCTACATGAAATCTGTCGCAGGCATTGG

GTGAGTGGGGACTGGAACTGGGCTGCATTGCTCATTTG  
AGAGATTANGTGCTCAGTGCTCCAGTGTTCCCAGAC  
TCCCCTGACATACCCAGGAAACAGGGCATGGGAAGGG  
AGAGGGTCCTATTGGGGTGGAAATCCAGTCCCTGCTGAT  
CTCTC

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## Figure 2F

### exon 10

ATGGCTCCTAAAGTGTTCAGCTCATTGTTATATTGG  
TGGTGAGGGTTAGTGTGTGAAAATTATACTAAACC  
TGTAGATGTTGATTCAGCAGAATTAGATCAAGTTT  
GGGTGTAAGACTTGTCCAACACCTATGTCTTGCTTAT  
TTCCAG

ACAAACTGGGAAGATTGAGCCTGTGGCCTGCCGCTGCT  
CTGGTTGCAGAG

GTAAGGGTGCCTGGCACAGCGTCGGGGCTTGT  
ATAGCCAATGTGGGCATTGAGGCAGGAGGCAGGG  
AGCACCTGTAGAAAGGGAGAGGGCTGAGCCAGGGTAAC  
CGGACTGTTACATGGACCAGCGTATCATACACTTCACCC  
TGTC

### exon 11

CCTGGAGGGAGGAGGTCCCTGGCAGGCTCCAACACATGC  
TTTAGCCGGAAAGCTTGAGGTGGGAAAGCTGAGGCAGG  
GCACAGAGGAAGGTGTTGGGTGGCATCTGCGCTGTAG  
CCCGCAGGCTGGGCCAGCTCATGTGTTGTCAATTCT  
GTCTCCTCAG

AGCGGGGCCATGGAGGGGGAGACTCTCACACATTCTAC  
ACTCAGCTGGTGTGATGCCAAGGTGATGCACATGCC  
CAGTACGTCCCTGGCGCTGGCTGCGTCCTGCTGCTG  
GTCCCTGTCATGCCAAATCCGGAGCAA

GTTAGGTGCTGGCCAGAGGGCAGCCCCGGCTGACAGCCAT  
TCGCTTGCCTGCTGGGGAAAGGGGCCAGATCGGACC  
CTCTGGCCAACCGCAGCCTGGAGCCACCTCCAGCAG  
CAGTCCTGCGTCTGCGGAGTGGGAGCGGTCACTGCT  
GGGGG

## Figure 2G

exon 12

CCCCACATCTCAGCCACCTGCAATCGTGAGGGTTGTTG  
GAECTAAACTTATGTGCCCTTCCTGTTCCCTTTGCC  
TTTGCAAATTGAAGAACCGTGTAAAACCATTTTAT  
GTGGCTTCAACGTCAACTATAAATTAGCTTGGTTATCTT  
CTAG

GAGAAATGCTATTTATTGGAGTAGTAGTAAAAAGGGC  
TCAAAGGATAAGGAGGCATTAGGCCTATTCTGAATCC  
CTGATGACATCAGCTCCAAGGGCTCTGTGCTGCAGGAA  
GCAAAACGTAG

GTGGGTACCAGGTAATGCCGTGCGCCTCCCCGCCCCCTC  
CCATATCAAGTAGAATGCTGGCGGCTTAAACATTGGG  
GTCCTGCTCATTCCTTCAGCCTCAACTTCACCTGGAG  
TGTCTACAGACTGAAGATGCATATTGTGTTGGCTT  
TTGGAGAAA

**Figure 3A**

Figure 3B

→ exon 9

F L D I H P V T G I P M N C S V K L Q L	390
TTC CTG GAC ATC CAC CCG GTC ACG GGA ATC CCC ATG AAC TGC TCT GTG AAA CTG CAG CTG	1288

→ exon 10

S L Y M K S V A G I G Q T G K I E P V V	410
AGC CTC TAC ATG AAA TCT GTC GCA GGC ATT GGA CAA ACT GGG AAG ATT GAG CCT GTG GTC	1348

→ exon 11

L P L L W F A E S G A M E G E T L H T F	430
CTG CCG CTG CTC TGG TTT GCA GAG AGC GGG GCC ATG GAG GGG GAG ACT CCT CAC ACA TTC	1408

→ exon 12

Y T Q L V L M P K V M H Y A Q Y V L L A	450
TAC ACT CAG CTG GTG TTG ATG CCC AAG GTG ATG CAC TAT GCC CAG TAC GTC CTC CTG GCG	1468

→ exon 13

L G C V L L L V P V I C Q I R S Q E K C	470
CTG GGC TGC CTG CTG GTC CCT GTC ATC TGC CAA ATC CGG AGC CAA GAG AAA TGC	1528

→ exon 13

Y L F W S S S K K G S K D K E A I Q A Y	490
TAT TTA TTT TGG AGT AGT AAA AAG GGC TCA AAG GAT AAG GAG GCC ATT CAG GCC TAT	1588

→ exon 13

S E S L M T S A P K G S V L Q E A K L *	510
TCT GAA TCC CTG ATG ACA TCA GCT CCC AAG GGC TCT GTG CTG CAG GAA GCA AAA CTG TAG	1643

GGTCCGTAGGACACCGTGAGCCAGCCAGGCCCTGGCGCTGGGCCTGACCGGCCCCCAGCCCCAACACCCCGCTTCTCC 1727

CGGACTCTCCCACCAAGACAGCCCCACAGGCTGAGCTCTCCAGCTGCCATGTGCCTGTTGCACACCTGCACA 1806

CACGCCCTGGCACACATACACACATGCGTGCAGGCTTGTGCAGACACTCAGGGATGGAGCTGCTGCTGAAGGGACTTGT 1855

AGGGAGAGGCTGTCAAACAAGCACTGTCTGGAACCTCTCTCCACGTGGCCCACAGGCCTGACCACAGGGCTGTGG 1964

TCCCTGGGTCCCCCTCTCGGGTGAGCCTGGCTTCCCATGCAGGACGGGCCAGGCTGGAGTCCCCCTTCTGTGCCAAATTCAAGT 2043

ACACTGCAAGTCCCCGGTGTGGTGGCTCCCCATGCAGGACGGGCCAGGCTGGAGTCCCCCTTCTGTGCCAAATTCAAGT 2122

GGGGACTCAGTGCCTAGGCCAGGCCCTGGCCACGAGCTTGGCTTGGTCTACCTGCCAGGCAGGCAAAGGCCCTTACACAG 2201

GCCTGGAAAAACAATGGAGTGAACACAAGATGCCCTGTGCAGCTGCCAGGGCTCOGCCACCCGGCCGGACTTTG 2230

ATCCCCCGAAGTCTCACAGGCACTGCATGGGTGTCTGGGCCCTTTCTCCATGCCCTAAACTGACATCATCCTAT 2359

GGACTGAGCCGGCCACTYYTGGCGAAGTGGCGCAGGCTGTGCCCCCGAGCTGCCCTTCCACAGGGTCCCT 2438

CAGATTATAGGTGCCCAAGGCTGAGGTGAAGAGGCCCTGGGGCCCTGCCCTCCGGGCGCTCCTGGACCCCTGGGCAAACC 2517

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AAAAAAAAAATGGGAAAAAAAAAAAAAAAAAAAAAA 2630

Figure 4

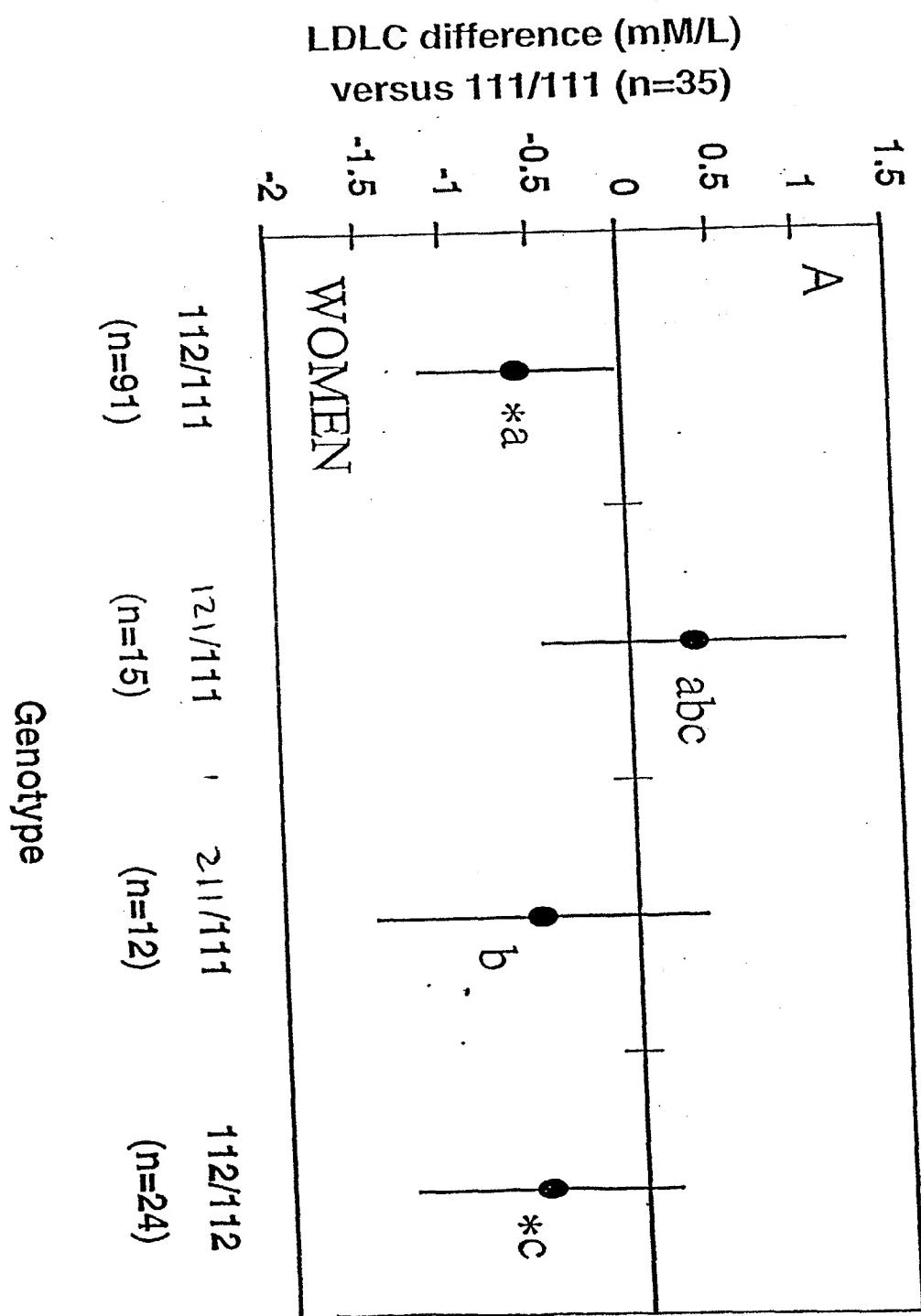


Figure 5

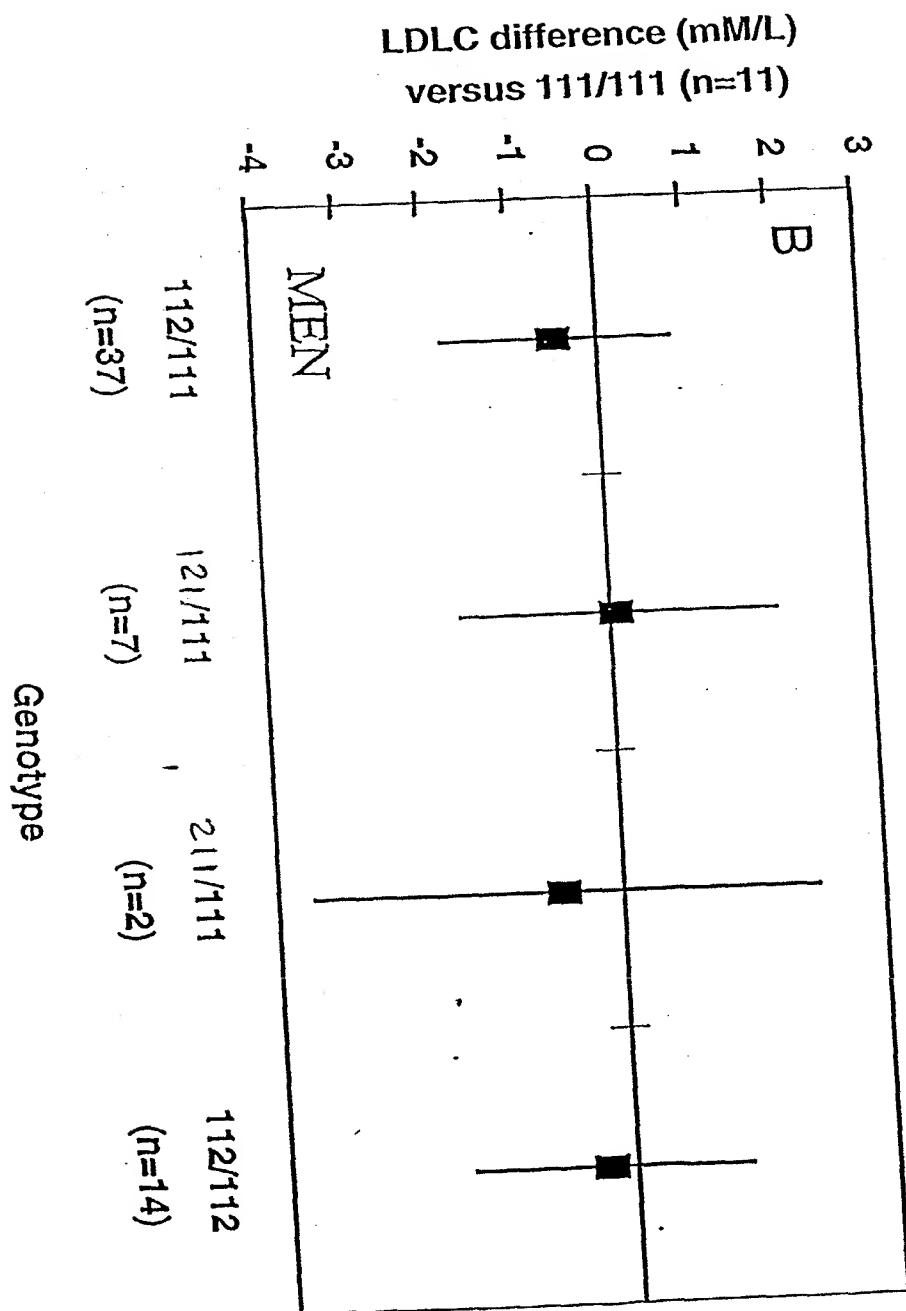


Figure 6

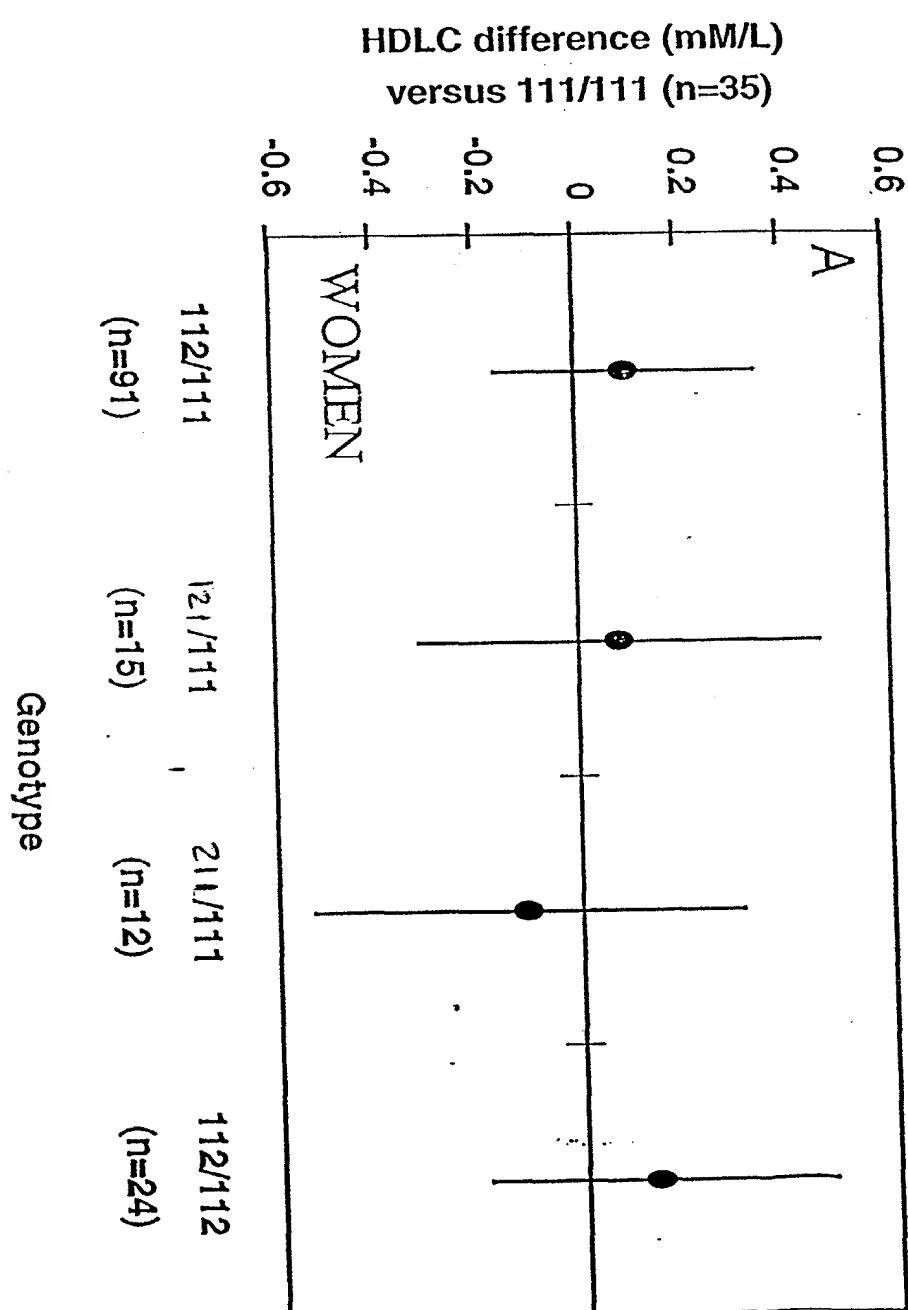


Figure 7

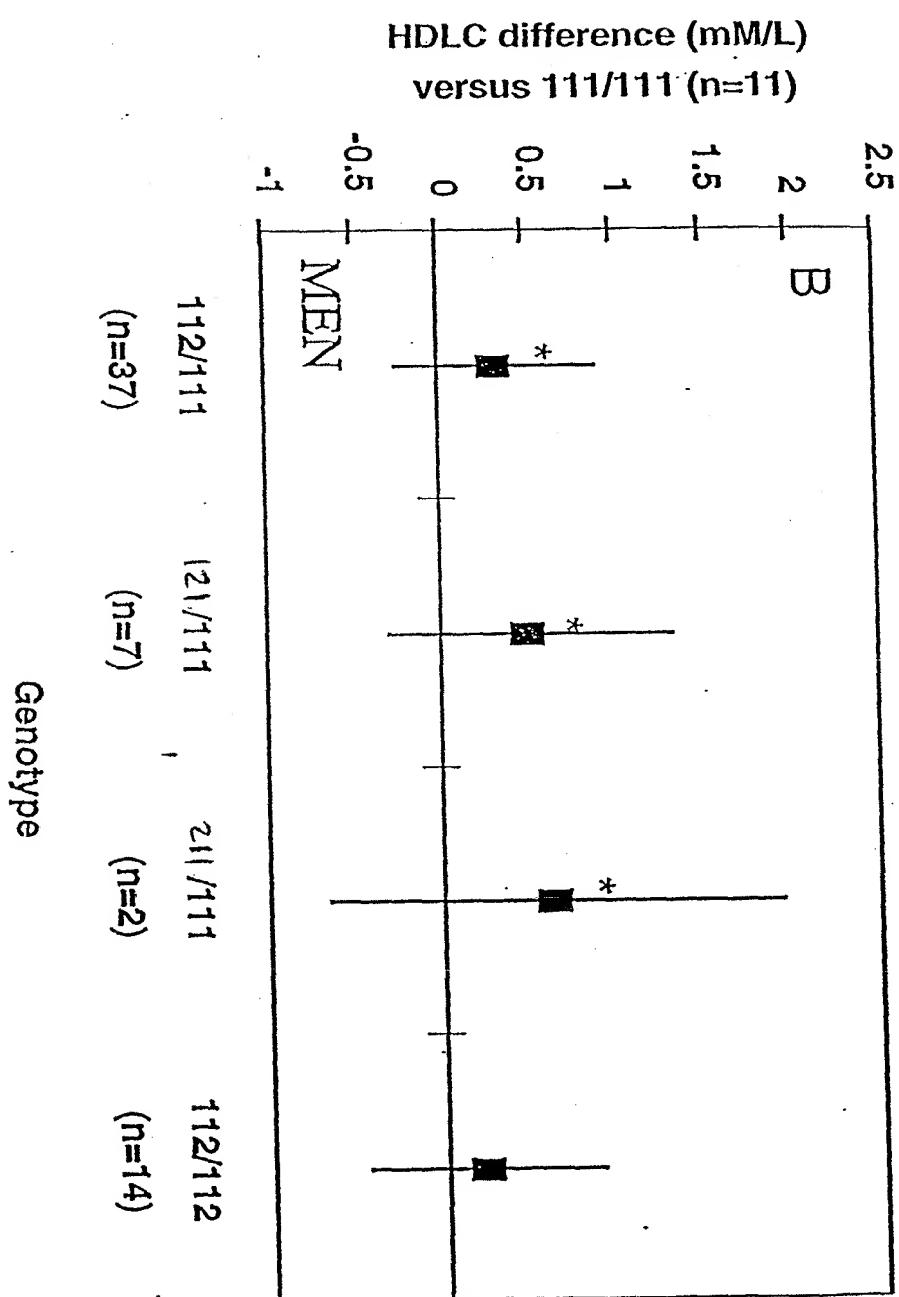


Figure 8

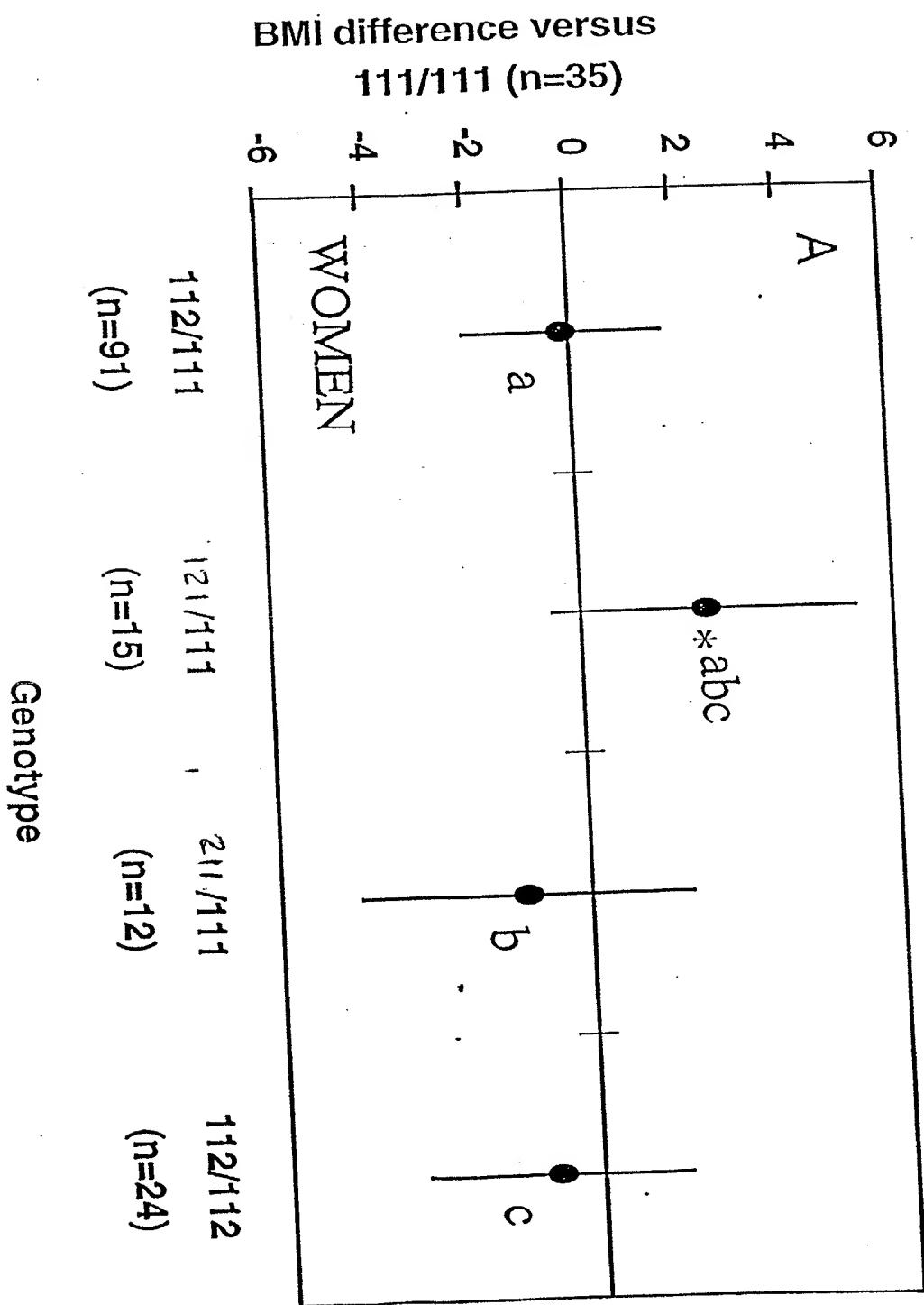


Figure 9

